



RPS10 gene

ribosomal protein S10

Normal Function

The *RPS10* gene provides instructions for making one of approximately 80 different ribosomal proteins, which are components of cellular structures called ribosomes. Ribosomes process the cell's genetic instructions to create proteins.

Each ribosome is made up of two parts (subunits) called the large and small subunits. The protein produced from the *RPS10* gene is among those found in the small subunit.

The specific functions of the RPS10 protein and the other ribosomal proteins within these subunits are unclear. Some ribosomal proteins are involved in the assembly or stability of ribosomes. Others help carry out the ribosome's main function of building new proteins. Studies suggest that some ribosomal proteins may have other functions, such as participating in chemical signaling pathways within the cell, regulating cell division, and controlling the self-destruction of cells (apoptosis).

Health Conditions Related to Genetic Changes

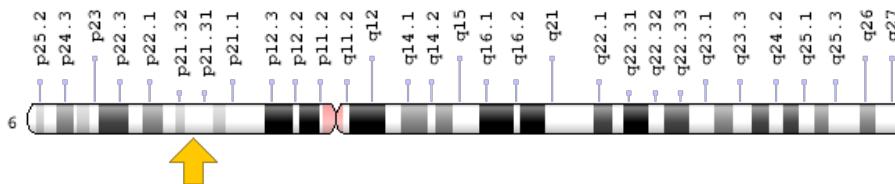
Diamond-Blackfan anemia

At least three *RPS10* gene mutations have been identified in individuals with Diamond-Blackfan anemia. These mutations are believed to result in an abnormally short, nonfunctional RPS10 protein that may impair the assembly of ribosomes, but the specific effects of the mutations are not known. Studies indicate that a shortage of functioning ribosomal proteins may increase the self-destruction of blood-forming cells in the bone marrow, resulting in a low number of red blood cells (anemia). Abnormal regulation of cell division or inappropriate triggering of apoptosis may contribute to the other health problems and unusual physical features that affect some people with Diamond-Blackfan anemia.

Chromosomal Location

Cytogenetic Location: 6p21.31, which is the short (p) arm of chromosome 6 at position 21.31

Molecular Location: base pairs 34,417,454 to 34,426,125 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 40S ribosomal protein S10
- DBA9
- MGC88819
- RS10_HUMAN
- S10

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The RNA message is decoded on ribosomes
<https://www.ncbi.nlm.nih.gov/books/NBK26829/#A1071>

GeneReviews

- Diamond-Blackfan Anemia
<https://www.ncbi.nlm.nih.gov/books/NBK7047>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RPS10%5BTIAB%5D%29+OR+%28ribosomal+protein+S10%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- RIBOSOMAL PROTEIN S10
<http://omim.org/entry/603632>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_RPS10.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RPS10%5Bgene%5D>
- Diamond-Blackfan Anemia Mutation Database
http://www.dbgenes.unito.it/home.php?select_db=RPS10
- HGNC Gene Family: S ribosomal proteins
<http://www.genenames.org/cgi-bin/genefamilies/set/728>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10383
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6204>
- UniProt
<http://www.uniprot.org/uniprot/P46783>

Sources for This Summary

- Boria I, Garelli E, Gazda HT, Aspesi A, Quarello P, Pavesi E, Ferrante D, Meerpohl JJ, Kartal M, Da Costa L, Proust A, Leblanc T, Simansour M, Dahl N, Fröjmark AS, Pospisilova D, Cmejla R, Beggs AH, Sheen MR, Landowski M, Buros CM, Clinton CM, Dobson LJ, Vlachos A, Atsidaftos E, Lipton JM, Ellis SR, Ramenghi U, Dianzani I. The ribosomal basis of Diamond-Blackfan Anemia: mutation and database update. *Hum Mutat.* 2010 Dec;31(12):1269-79. doi: 10.1002/humu.21383.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20960466>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4485435/>
- Doherty L, Sheen MR, Vlachos A, Choesmel V, O'Donohue MF, Clinton C, Schneider HE, Sieff CA, Newburger PE, Ball SE, Niewiadomska E, Matysiak M, Glader B, Arceci RJ, Farrar JE, Atsidaftos E, Lipton JM, Gleizes PE, Gazda HT. Ribosomal protein genes RPS10 and RPS26 are commonly mutated in Diamond-Blackfan anemia. *Am J Hum Genet.* 2010 Feb 12;86(2):222-8. doi: 10.1016/j.ajhg.2009.12.015. Epub 2010 Jan 28. Erratum in: *Am J Hum Genet.* 2010 Apr 9;86(4):655.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20116044>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2820177/>
- Ellis SR, Gleizes PE. Diamond Blackfan anemia: ribosomal proteins going rogue. *Semin Hematol.* 2011 Apr;48(2):89-96. doi: 10.1053/j.seminhematol.2011.02.005. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21435505>
- Farrar JE, Dahl N. Untangling the phenotypic heterogeneity of Diamond Blackfan anemia. *Semin Hematol.* 2011 Apr;48(2):124-35. doi: 10.1053/j.seminhematol.2011.02.003. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21435509>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3078697/>
- Ito E, Konno Y, Toki T, Terui K. Molecular pathogenesis in Diamond-Blackfan anemia. *Int J Hematol.* 2010 Oct;92(3):413-8. doi: 10.1007/s12185-010-0693-7. Epub 2010 Sep 30. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20882441>
- Narla A, Hurst SN, Ebert BL. Ribosome defects in disorders of erythropoiesis. *Int J Hematol.* 2011 Feb;93(2):144-9. doi: 10.1007/s12185-011-0776-0. Epub 2011 Feb 1. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21279816>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3689295/>
- OMIM: RIBOSOMAL PROTEIN S10
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